HNRNPK gene

heterogeneous nuclear ribonucleoprotein K

Normal Function

The *HNRNPK* gene provides instructions for making a protein called heterogenous nuclear ribonucleoprotein K (hnRNP K). This protein attaches (binds) to DNA or its chemical cousin RNA and to other proteins. It acts as a docking site to bring together different molecules in the cell, which is important for relaying signals and controlling cellular functions. By bringing certain proteins together with DNA or RNA, the hnRNP K protein helps control the activity of genes and the production of proteins. By regulating gene activity and protein production, hnRNP K is involved in many cellular processes, including growth and division (proliferation) of cells, maturation of cells to take on specialized function (differentiation), and self-destruction (apoptosis) of cells when they are no longer needed.

The hnRNP K protein plays a role in the normal development or function of many body systems. In the brain, the protein may be involved in a process called synaptic plasticity, which is the ability of the connections between neurons (synapses) to change and adapt over time in response to experience. This process is critical for learning and memory. Another process in the brain involving the hnRNP K protein is the growth of nerve cell extensions called axons, which are essential for transmission of nerve impulses. While the protein is likely critical in other systems and processes, its role is not well understood.

Health Conditions Related to Genetic Changes

Au-Kline syndrome

More than 20 mutations in the *HNRNPK* gene have been found to cause Au-Kline syndrome. This condition is characterized by weak muscle tone (hypotonia), intellectual disability, delayed development of speech and walking, and distinctive facial features. Other body systems, such as the heart, kidneys, and bones, may also be affected.

The *HNRNPK* gene mutations that cause Au-Kline syndrome alter the blueprint for making hnRNP K protein, which results in the production of little or no hnRNP K protein from one copy of the gene. A shortage of this protein changes gene activity and protein production in cells. As a result, the normal development or function of cells, tissues, and organs in many parts of the body is disrupted, leading to the varied features of Au-Kline syndrome. In particular, problems with brain development likely contribute to intellectual disability, delayed development, and other neurological problems in people with the condition.

Cancers

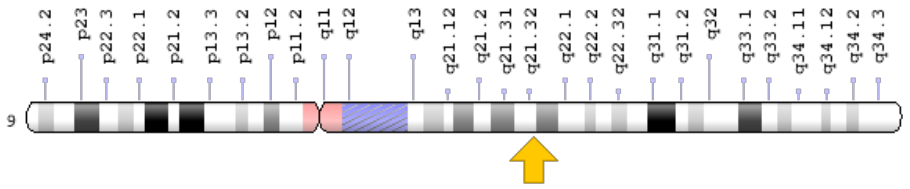
Loss of the *HNRNPK* gene is thought to be involved in the development of a blood cancer known as acute myeloid leukemia (AML). A mutation that removes (deletes) DNA from the long (q) arm of chromosome 9 occurs in about 2 percent of AML cases. Such cases are referred to as del(9q) AML. The missing region of DNA includes the *HNRNPK* gene, among other genes. Researchers suspect that a shortage of hnRNP K protein alters the activity of genes that control cell growth, resulting in an excess of abnormal cells. When the del(9q) mutation occurs in certain blood cells, the shortage of hnRNP K may contribute to the uncontrolled cell growth that underlies AML.

Individuals with Au-Kline syndrome (described above) do not appear to have a higher risk of developing AML than does the general population.

Chromosomal Location

Cytogenetic Location: 9q21.32, which is the long (q) arm of chromosome 9 at position 21.32

Molecular Location: base pairs 83,968,083 to 83,980,782 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CSBP
- HNRPK
- transformation upregulated nuclear protein
- TUNP

Additional Information & Resources

Clinical Information from GeneReviews

- Au-Kline Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK540283

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HNRNPK%5BTIAB%5D%29+OR+%28heterogeneous+nuclear+ribonucleoprotein+K%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN K
  http://omim.org/entry/600712

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/HNRNPKID44314ch9q21.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HNRNPK%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:5044
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3190
- NCBI Gene
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